Table 1. Human circadian gene mutation and associated phenotypic effects/disorders.

Sleep Disorders			
Gene	Mutation	Phenotype/Disorder	Ref.
PER2	S662G[1]	FASPS	[1]
CK1δ	T44A[2]	FASPS	[2]
Gene	Seq. Variant	Phenotype/Disorder	Ref.
CLOCK	T3111C[2]	Diurnal preference, DSPS	[3-6]
	SNP rs10520010	Heritability of sleepiness	[7]
CK1ε	S408N[1]	Reduced susceptibility to DSPS/N-24	[8]
CK2A2	SNP rs28168	Heritability of bedtime	[7]
PER2	C111G[2]	Morning preference, ASPS	[9]
PER3	V647G[1]	DSPS	[10]
PER3	VNTR[1]	DSPS, Sleep homeostasis	[11-13]
PK2	SNP rs6599077	Heritability of sleep duration	[7]

Mood/Behavioral Disorders			
Gene	Seq. variant	Phenotype	Ref.
ARNTL/BMAL1	SNPs rs3789327 rs2278749	Associated with bipolar disorder	[14]
NPAS2	L471S[1]	Diurnal preference/SAD	[15]

PER3	SNPs rs228729 rs228642 rs228666 rs2859388 rs228697	Associated with bipolar disorder	[14]
ASMT (HIOMT)	SNPs rs4446909 rs5989681	Autism Spectrum Disorder	[16]

Metabolic Disorders			
Gene	Seq. variant	Phenotype	Ref.
BMAL1	SNPs rs7950226 rs6486121	Type 2 Diabetes Hypertension	[17]
CLOCK	SNPs rs486454 rs1801260	Metabolic Syndrome & Obesity	[18]

1. Amino acid substitution/variant.
2. Nucleotide substitution/variant.

ASPS: Advanced Sleep Phase Syndrome
FASPS: Familial Advanced Sleep Phase Syndrome
DSPS: Delayed Sleep Phase Syndrome
N-24: Non-24-hour Sleep-Wake Syndrome
SAD: Seasonal Affective Disorder
VNTR: Variable-Number Tandem-Repeat polymorphism

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